Alain Verloes

List of Publications by Year
in descending order

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Version: 2024-02-01

411 papers	20,953 citations	12597 71 h-index	18944 123 g-index
436 all docs	436 docs citations	436 times ranked	24312 citing authors

#	Article	IF	Citations
1	Neurological outcome in WDR62 primary microcephaly. Developmental Medicine and Child Neurology, 2022, 64, 509-517.	1.1	3
2	Management of growth failure and other endocrine aspects in patients with Noonan syndrome across Europe: A sub-analysis of a European clinical practice survey. European Journal of Medical Genetics, 2022, 65, 104404.	0.7	6
3	Rare and de novo duplications containing <scp>TCF20</scp> are associated with a neurodevelopmental disorder. Clinical Genetics, 2022, 101, 364-370.	1.0	7
4	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	1.1	32
5	Confirmation of FZD5 implication in a cohort of 50 patients with ocular coloboma. European Journal of Human Genetics, 2021, 29, 131-140.	1.4	10
6	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. European Journal of Human Genetics, 2021, 29, 524-527.	1.4	7
7	<scp>Smithâ€Magenis</scp> syndrome: Clinical and behavioral characteristics in a large retrospective cohort. Clinical Genetics, 2021, 99, 519-528.	1.0	14
8	Novel missense mutations in PTCHD1 alter its plasma membrane subcellular localization and cause intellectual disability and autism spectrum disorder. Human Mutation, 2021, 42, 848-861.	1.1	8
9	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
10	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	1.4	7
11	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
12	High rate of hypomorphic variants as the cause of inherited ataxia and related diseases: study of a cohort of 366 families. Genetics in Medicine, 2021, 23, 2160-2170.	1.1	13
13	<scp>EPHA7</scp> haploinsufficiency is associated with a neurodevelopmental disorder. Clinical Genetics, 2021, 100, 396-404.	1.0	3
14	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	0.6	6
15	10Âyears of CEMARA database in the AnDDI-Rares network: a unique resource facilitating research and epidemiology in developmental disorders in France. Orphanet Journal of Rare Diseases, 2021, 16, 345.	1.2	4
16	NTRK1 gene-related congenital insensitivity to pain with anhidrosis: a nationwide multicenter retrospective study. Neurogenetics, 2021, 22, 333-341.	0.7	5
17	Care management in a French cohort with Down syndrome from the AnDDI-Rares/CNSA study. European Journal of Medical Genetics, 2021, 64, 104290.	0.7	2
18	Oligo-astrocytoma in LZTR1-related Noonan syndrome. European Journal of Medical Genetics, 2020, 63, 103617.	0.7	17

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19	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	1.1	30
20	Digenic inheritance of human primary microcephaly delineates centrosomal and nonâ€centrosomal pathways. Human Mutation, 2020, 41, 512-524.	1.1	19
21	Giant axonal neuropathy: a multicenter retrospective study with genotypic spectrum expansion. Neurogenetics, 2020, 21, 29-37.	0.7	9
22	Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome. European Journal of Medical Genetics, 2020, 63, 104064.	0.7	5
23	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
24	<i>CDK5RAP2</i> primary microcephaly is associated with hypothalamic, retinal and cochlear developmental defects. Journal of Medical Genetics, 2020, 57, 389-399.	1.5	17
25	Report of the first patient with a homozygous <i>OTUD7A</i> variant responsible for epileptic encephalopathy and related proteasome dysfunction. Clinical Genetics, 2020, 97, 567-575.	1.0	18
26	Le syndrome ADNP (protÃ©ine neuroprotectrice dÃ©pendante de lâ€™activitÃ©) liÃ© Ã la dÃ©ficience intellectuelle et aux troubles du spectre autistiqueÂ: une revue de la littÃ©rature. Neuropsychiatrie De L'Enfance Et De L'Adolescence, 2020, 68, 93-99.	0.1	0
27	<i>LEF1</i> haploinsufficiency causes ectodermal dysplasia. Clinical Genetics, 2020, 97, 595-600.	1.0	11
28	Overlapping phenotypes between SHORT and Noonan syndromes in patients with PTPN11 pathogenic variants. Clinical Genetics, 2020, 98, 10-18.	1.0	9
29	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. Orphanet Journal of Rare Diseases, 2020, 15, 103.	1.2	23
30	Further delineation of the female phenotype with <scp><i>KDM5C</i></scp> disease causing variants: 19 new individuals and review of the literature. Clinical Genetics, 2020, 98, 43-55.	1.0	28
31	Dermatological manifestations in cardiofaciocutaneous syndrome: a prospective multicentric study of 45 mutationâ€positive patients. British Journal of Dermatology, 2019, 180, 172-180.	1.4	15
32	VPS51 biallelic variants cause microcephaly with brain malformations: A confirmatory report. European Journal of Medical Genetics, 2019, 62, 103704.	0.7	15
33	å¿ƒè„‚çš®è‚‚ç»¼åˆç—‡ä¸çš„çš®è‚¤è¡¨çŽ°. British Journal of Dermatology, 2019, 180, e30-e30.	1.4	0
34	åŠªå—ç»¼åˆå¾çš„çš®è‚‚ç—…è¡¨çŽ°. British Journal of Dermatology, 2019, 180, e266.	1.4	0
35	Dermatological manifestations in cardiofaciocutaneous syndrome. British Journal of Dermatology, 2019, 180, e17-e17.	1.4	0
36	Molecular investigation, using chromosomal microarray and whole exome sequencing, of six patients affected by Williams Beuren syndrome and Autism Spectrum Disorder. Orphanet Journal of Rare Diseases, 2019, 14, 121.	1.2	5

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37	Developmental Phenotype of the Rare Case of DJ Caused by a Unique ADNP Gene De Novo Mutation. Journal of Molecular Neuroscience, 2019, 68, 321-330.	1.1	21
38	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	1.1	58
39	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1223-1232.	2.6	43
40	Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. Genetics in Medicine, 2019, 21, 2043-2058.	1.1	57
41	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. Journal of Medical Genetics, 2019, 56, 526-535.	1.5	46
42	Dermatological manifestations in Noonan syndrome: aÂprospective multicentric study of 129 patients positive for mutation. British Journal of Dermatology, 2019, 180, 1438-1448.	1.4	20
43	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	0.7	24
44	Duplication of 10q24 locus: broadening the clinical and radiological spectrum. European Journal of Human Genetics, 2019, 27, 525-534.	1.4	13
45	INTUâ€related oralâ€facialâ€digital syndrome type VI: A confirmatory report. Clinical Genetics, 2018, 93, 1205-1209.	1.0	7
46	Diagnostic strategy in segmentation defect of the vertebrae: a retrospective study of 73 patients. Journal of Medical Genetics, 2018, 55, 422.2-429.	1.5	14
47	EFNB2 haploinsufficiency causes a syndromic neurodevelopmental disorder. Clinical Genetics, 2018, 93, 1141-1147.	1.0	18
48	MED13L-related intellectual disability: involvement of missense variants and delineation of the phenotype. Neurogenetics, 2018, 19, 93-103.	0.7	25
49	Disease-causing variants in TCF4 are a frequent cause of intellectual disability: lessons from large-scale sequencing approaches in diagnosis. European Journal of Human Genetics, 2018, 26, 996-1006.	1.4	13
50	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661.	3.7	52
51	Down syndrome-like acute megakaryoblastic leukemia in a patient with Cornelia de Lange syndrome. Haematologica, 2018, 103, e274-e276.	1.7	9
52	Incidence of infantile Pompe disease in the Maroon population of French Guiana. BMJ Paediatrics Open, 2018, 2, e000182.	0.6	14
53	Autosomal recessive primary microcephaly due to ASPM mutations: An update. Human Mutation, 2018, 39, 319-332.	1.1	53
54	Golgipathies in Neurodevelopment: A New View of Old Defects. Developmental Neuroscience, 2018, 40, 396-416.	1.0	35

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55	NR4A2 haploinsufficiency is associated with intellectual disability and autism spectrum disorder. Clinical Genetics, 2018, 94, 264-268.	1.0	22
56	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50.	3.7	1
57	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	1.4	18
58	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. European Journal of Human Genetics, 2018, 26, 64-74.	1.4	72
59	Noonan syndrome males display Sertoli cell-specific primary testicular insufficiency. European Journal of Endocrinology, 2018, 179, 409-418.	1.9	16
60	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
61	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	1.1	90
62	Phosphoglycerate dehydrogenase (PHGDH) deficiency without epilepsy mimicking primary microcephaly. American Journal of Medical Genetics, Part A, 2017, 173, 1936-1942.	0.7	8
63	Molecular and clinical delineation of 2p15p16.1 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2081-2087.	0.7	18
64	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	1.8	66
65	<scp>DNA</scp> ligase <scp>IV</scp> deficiency: Immunoglobulin class deficiency depends on the genotype. Pediatric Allergy and Immunology, 2017, 28, 298-303.	1.1	7
66	Facial dysmorphism is influenced by ethnic background of the patient and of the evaluator. Clinical Genetics, 2017, 92, 166-171.	1.0	63
67	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
68	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. Brain, 2017, 140, 2597-2609.	3.7	28
69	Identification of STAC3 variants in nonâ€Native American families with overlapping features of Careyâ€“Finemanâ€“Ziter syndrome and Moebius syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2763-2771.	0.7	28
70	Autosomal recessive variations of <scp>TBX6</scp>, from congenital scoliosis to spondylocostal dysostosis. Clinical Genetics, 2017, 91, 908-912.	1.0	38
71	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	1.7	58
72	Infantile systemic hyalinosis: a report of two new cases, one with prolonged survival. European Journal of Dermatology, 2017, 27, 328-329.	0.3	2

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73	CHARGE and Kabuki Syndromes: Gene-Specific DNA Methylation Signatures Identify Epigenetic Mechanisms Linking These Clinically Overlapping Conditions. American Journal of Human Genetics, 2017, 100, 773-788.	2.6	166
74	Fetal phenotypes in otopalatodigital spectrum disorders. Clinical Genetics, 2016, 89, 371-377.	1.0	17
75	Refining the phenotypical and mutational spectrum of Taybiâ€Linder syndrome. Clinical Genetics, 2016, 90, 550-555.	1.0	14
76	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	1.1	134
77	Prenatal findings in cardioâ€facioâ€cutaneous syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 441-445.	0.7	10
78	Fraser syndrome: features suggestive of prenatal diagnosis in a review of 38 cases. Prenatal Diagnosis, 2016, 36, 1270-1275.	1.1	25
79	Incomplete penetrance of biallelic ALDH1A3 mutations. European Journal of Medical Genetics, 2016, 59, 215-218.	0.7	13
80	Failure of ossification of the occipital bone in mandibuloacral dysplasia type B. American Journal of Medical Genetics, Part A, 2016, 170, 2750-2755.	0.7	12
81	ARCN1 Mutations Cause a Recognizable Craniofacial Syndrome Due to COPI-Mediated Transport Defects. American Journal of Human Genetics, 2016, 99, 451-459.	2.6	65
82	ARID1Bmutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. Brain, 2016, 139, e64-e64.	3.7	26
83	Mutations in Citron Kinase Cause Recessive Microlissencephaly with Multinucleated Neurons. American Journal of Human Genetics, 2016, 99, 511-520.	2.6	59
84	Copy number variants and rasopathies: germline KRAS duplication in a patient with syndrome including pigmentation abnormalities. Orphanet Journal of Rare Diseases, 2016, 11, 101.	1.2	7
85	Mutations in RIT1 cause Noonan syndrome with possible juvenile myelomonocytic leukemia but are not involved in acute lymphoblastic leukemia. European Journal of Human Genetics, 2016, 24, 1124-1131.	1.4	23
86	Acute lymphoblastic leukemia in the context of RASopathies. European Journal of Medical Genetics, 2016, 59, 173-178.	0.7	35
87	Growth patterns of patients with Noonan syndrome: correlation with age and genotype. European Journal of Endocrinology, 2016, 174, 641-650.	1.9	40
88	Abnormal spindle-like microcephaly-associated (ASPM) mutations strongly disrupt neocortical structure but spare the hippocampus and long-term memory. Cortex, 2016, 74, 158-176.	1.1	32
89	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	1.1	125
90	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67

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91	11q24.2â€25 microâ€rearrangements in autism spectrum disorders: Relation to brain structures. American Journal of Medical Genetics, Part A, 2015, 167, 3019-3030.	0.7	25
92	Mutations in the latent TGF-beta binding protein 3 (LTBP3) gene cause brachyolmia with amelogenesis imperfecta. Human Molecular Genetics, 2015, 24, 3038-3049.	1.4	40
93	Variable expression pattern in Donnai-Barrow syndrome: Report of two novel LRP2 mutations and review of the literature. European Journal of Medical Genetics, 2015, 58, 293-299.	0.7	55
94	Inherited 1q21.1q21.2 duplication and 16p11.2 deletion: A two-hit case with more severe clinical manifestations. European Journal of Medical Genetics, 2015, 58, 497-501.	0.7	7
95	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. Molecular Autism, 2015, 6, 19.	2.6	29
96	Clinical and molecular delineation of Tetrasomy 9p syndrome: Report of 12 new cases and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1252-1261.	0.7	20
97	Mutations in TUBGCP4 Alter Microtubule Organization via the Î³-Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. American Journal of Human Genetics, 2015, 96, 666-674.	2.6	60
98	Clinical utility gene card for: CHARGE syndrome - update 2015. European Journal of Human Genetics, 2015, 23, 3-4.	1.4	18
99	Mutations of the ImprintedCDKN1CGene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. Human Mutation, 2015, 36, 894-902.	1.1	62
100	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. European Journal of Human Genetics, 2015, 23, 1010-1018.	1.4	35
101	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. Nature Genetics, 2015, 47, 73-77.	9.4	130
102	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	1.4	115
103	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	1.4	38
104	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	1.1	50
105	Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome. American Journal of Human Genetics, 2014, 95, 637-648.	2.6	108
106	Neonatal progeroid variant of Marfan syndrome with congenital lipodystrophy results from mutations at the 3â€² end of FBN1 gene. European Journal of Medical Genetics, 2014, 57, 230-234.	0.7	41
107	Large deletions encompassing the TCOF1 and CAMK2A genes are responsible for Treacher Collins syndrome with intellectual disability. European Journal of Human Genetics, 2014, 22, 52-56.	1.4	22
108	Duplication of the 15q11-q13 region: Clinical and genetic study of 30 new cases. European Journal of Medical Genetics, 2014, 57, 5-14.	0.7	68

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109	Severe forms of Baraitserâ€“Winter syndrome are caused by ACTB mutations rather than ACTG1 mutations. European Journal of Human Genetics, 2014, 22, 179-183.	1.4	67
110	Simplified gyral pattern in severe developmental microcephalies? New insights from allometric modeling for spatial and spectral analysis of gyrification. NeuroImage, 2014, 102, 317-331.	2.1	32
111	Juvenile myelomonocytic leukaemia and Noonan syndrome. Journal of Medical Genetics, 2014, 51, 689-697.	1.5	112
112	Blepharophimosis, short humeri, developmental delay and hirschsprung disease: Expanding the phenotypic spectrum of MED12 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 1821-1825.	0.7	19
113	Cerebro-fronto-facial syndrome type 3 with polymicrogyria: A clinical presentation of Baraitserâ€“Winter syndrome. European Journal of Medical Genetics, 2014, 57, 32-36.	0.7	14
114	Heterozygous Loss-of-Function Mutations in YAP1 Cause Both Isolated and Syndromic Optic Fissure Closure Defects. American Journal of Human Genetics, 2014, 94, 295-302.	2.6	93
115	Identification of Nine New RAI1-Truncating Mutations in Smith-Magenis Syndrome Patients without 17p11.2 Deletions. Molecular Syndromology, 2014, 5, 57-64.	0.3	22
116	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.5	84
117	The development of a clinical screening tool for tumour predisposition syndromes in childhood cancer patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 52-52.	0.0	0
118	Microcephaly. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 129-141.	1.0	94
119	The development of a clinical screening instrument for tumour predisposition syndromes in childhood cancer patients. European Journal of Cancer, 2013, 49, 3247-3254.	1.3	18
120	Presence of autism, hyperserotonemia, and severe expressive language impairment in Williams-Beuren syndrome. Molecular Autism, 2013, 4, 29.	2.6	33
121	A de novo 17q21.2 duplication in a boy with developmental delay and dysmorphic features. European Journal of Medical Genetics, 2013, 56, 226-228.	0.7	1
122	Mutations in WNT10A are frequently involved in oligodontia associated with minor signs of ectodermal dysplasia. American Journal of Medical Genetics, Part A, 2013, 161, 671-678.	0.7	52
123	Phenotypic Spectrum of Simpsonâ€“<scp>G</scp>olabiâ€“<scp>B</scp>ehmel Syndrome in a Series of 42 Cases With a Mutation in <scp>GPC</scp>3 and Review of the Literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 92-105.	0.7	78
124	Clinical utility gene card for: Rothmundâ€“Thomson syndrome. European Journal of Human Genetics, 2013, 21, 792-792.	1.4	18
125	Beckwithâ€“Wiedemann syndrome and long <scp>QT</scp> syndrome due to familialâ€balanced translocation t(11;17)(p15.5;q21.3) involving the <scp>KCNQ1</scp> gene. Clinical Genetics, 2013, 84, 78-81.	1.0	18
126	Reduced dosage of ERF causes complex craniosynostosis in humans and mice and links ERK1/2 signaling to regulation of osteogenesis. Nature Genetics, 2013, 45, 308-313.	9.4	141

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127	Developmental anomalies of the lids. , 2013, , 147-164.e3.		3
128	Genomic imbalances detected by array-CGH in patients with syndromal ocular developmental anomalies. European Journal of Human Genetics, 2012, 20, 527-533.	1.4	19
129	Autism multiplex family with 16p11.2p12.2 microduplication syndrome in monozygotic twins and distal 16p11.2 deletion in their brother. European Journal of Human Genetics, 2012, 20, 540-546.	1.4	38
130	Mutations at a single codon in Mad homology 2 domain of SMAD4 cause Myhre syndrome. Nature Genetics, 2012, 44, 85-88.	9.4	125
131	Targeted high-throughput sequencing for diagnosis of genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and AlstrÃ¶m Syndromes. Journal of Medical Genetics, 2012, 49, 502-512.	1.5	104
132	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	9.4	237
133	Mutation of the parkinsonism gene ATP13A2 causes neuronal ceroid-lipofuscinosis. Human Molecular Genetics, 2012, 21, 2646-2650.	1.4	231
134	Phenotypic spectrum of fetal Smithâ€“Lemliâ€“Opitz syndrome. European Journal of Medical Genetics, 2012, 55, 81-90.	0.7	32
135	A new lysosomal storage disorder resembling Morquio syndrome in sibs. European Journal of Medical Genetics, 2012, 55, 157-162.	0.7	2
136	Constitutional NRAS mutations are rare among patients with Noonan syndrome or juvenile myelomonocytic leukemia. American Journal of Medical Genetics, Part A, 2012, 158A, 2407-2411.	0.7	20
137	Preâ€•and postnatal phenotype of 6p25 deletions involving the FOXC1 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 2430-2438.	0.7	30
138	Mutations in the Î²-Tubulin Gene TUBB5 Cause Microcephaly with Structural Brain Abnormalities. Cell Reports, 2012, 2, 1554-1562.	2.9	162
139	Exome sequencing identifies mutations inLZTFL1, a BBSome and smoothened trafficking regulator, in a family with Bardetâ€“Biedl syndrome with situs inversus and insertional polydactyly. Journal of Medical Genetics, 2012, 49, 317-321.	1.5	119
140	Pseudoaminopterin syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2233-2238.	0.7	0
141	Extensive abdominal lipomatosis in a patient with Noonan/LEOPARD syndrome (Noonan) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50	0.7	1
142	Nail and phalangeal agenesis in a patient with 4pter and 9pter duplication. American Journal of Medical Genetics, Part A, 2012, 158A, 2277-2282.	0.7	3
143	Haploinsufficiency ofSOX5at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. Human Mutation, 2012, 33, 728-740.	1.1	85
144	Autistic Disorder in Patients with Williams-Beuren Syndrome: A Reconsideration of the Williams-Beuren Syndrome Phenotype. PLoS ONE, 2012, 7, e30778.	1.1	46

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145	Molecular diagnosis reveals genetic heterogeneity for the overlapping MKKS and BBS phenotypes. European Journal of Medical Genetics, 2011, 54, 157-160.	0.7	39
146	Terminal 4q deletion and 8q duplication in a patient with CHARGE-like features. European Journal of Medical Genetics, 2011, 54, 173-176.	0.7	8
147	A familial syndromal form of omphalocele. European Journal of Medical Genetics, 2011, 54, 337-340.	0.7	3
148	Hippocampal dysgenesis and variable neuropsychiatric phenotypes in patients with Bardet-Biedl syndrome underline complex CNS impact of primary cilia. Clinical Genetics, 2011, 80, 523-531.	1.0	32
149	Clinical utility gene card for: WAGR syndrome. European Journal of Human Genetics, 2011, 19, 492-492.	1.4	24
150	Dissection of the MYCN locus in Feingold syndrome and isolated oesophageal atresia. European Journal of Human Genetics, 2011, 19, 602-606.	1.4	24
151	Clinical utility gene card for: CHARGE syndrome. European Journal of Human Genetics, 2011, 19, 1017-1017.	1.4	6
152	Dyggveâ€“Melchiorâ€“Clausen syndrome: novel splice mutation with atlanto-axial subluxation. European Journal of Pediatrics, 2011, 170, 121-126.	1.3	6
153	Cardioâ€facioâ€cutaneous syndrome: Does genotype predict phenotype?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 129-135.	0.7	72
154	A Long-term Competent Chimeric Immune System in a Dizygotic Dichorionic Twin. Pediatrics, 2011, 128, e458-e463.	1.0	9
155	Aphallia, Lung Agenesis and Multiple Defects of Blastogenesis. Fetal and Pediatric Pathology, 2011, 30, 22-26.	0.4	9
156	VIP blockade leads to microcephaly in mice via disruption of Mcph1-Chk1 signaling. Journal of Clinical Investigation, 2011, 121, 3072-3087.	3.9	23
157	Mesomelia-Synostoses Syndrome Results from Deletion of SULF1 and SLCO5A1 Genes at 8q13. American Journal of Human Genetics, 2010, 87, 95-100.	2.6	42
158	Identification of 28 novel mutations in the Bardetâ€“Biedl syndrome genes: the burden of private mutations in an extensively heterogeneous disease. Human Genetics, 2010, 127, 583-593.	1.8	109
159	The RIN2 syndrome: a new autosomal recessive connective tissue disorder caused by deficiency of Ras and Rab interactor 2 (RIN2). Human Genetics, 2010, 128, 79-88.	1.8	51
160	Molecular and clinical analysis ofRAF1in Noonan syndrome and related disorders: dephosphorylation of serine 259 as the essential mechanism for mutant activation. Human Mutation, 2010, 31, 284-294.	1.1	96
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